



sudden infant death with dysgenesis of the testes syndrome

Sudden infant death with dysgenesis of the testes syndrome (SIDDT) is a rare condition that is fatal in the first year of life; its major features include abnormalities of the reproductive system in males, feeding difficulties, and breathing problems.

Infants with SIDDT who are genetically male, with one X chromosome and one Y chromosome in each cell, have underdeveloped or abnormal testes. They may also have external genitalia that appear female or that do not look clearly male or clearly female (ambiguous genitalia). In affected infants who are genetically female, with two X chromosomes in each cell, development of the internal and external reproductive organs is normal.

SIDDT is associated with abnormal development of the brain, particularly the brainstem, which is the part of the brain that is connected to the spinal cord. The brainstem regulates many basic body functions, including heart rate, breathing, eating, and sleeping. It also relays information about movement and the senses between the brain and the rest of the body. Many features of SIDDT appear to be related to brainstem malfunction, including a slow or uneven heart rate, abnormal breathing patterns, difficulty controlling body temperature, unusual tongue and eye movements, abnormal reflexes, seizures, and feeding difficulties. Affected infants also have an unusual cry that has been described as similar to the bleating of a goat, which is probably a result of abnormal nerve connections between the brain and the voicebox (larynx).

The brainstem abnormalities lead to death in the first year of life, when affected infants suddenly stop breathing or their heart stops beating (cardiorespiratory arrest).

Frequency

SIDDT has been diagnosed in more than 20 infants from a single Old Order Amish community in Pennsylvania. The condition has not been reported outside this community.

Genetic Changes

A single mutation in the *TSPYL1* gene has caused all identified cases of SIDDT. This gene provides instructions for making a protein called TSPY-like 1, whose function is unknown. Based on its role in SIDDT, researchers propose that TSPY-like 1 is involved in the development of the male reproductive system and the brain.

The *TSPYL1* gene mutation that causes SIDDT eliminates the function of TSPY-like 1. The loss of this protein's function appears to cause the major features of the disorder.

by disrupting the normal development of the male reproductive system and the brain, particularly the brainstem.

Research findings suggest that mutations in the *TSPYL1* gene are not associated with sudden infant death syndrome (SIDS) in the general population. SIDS is a major cause of death in children younger than 1 year.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- SIDDT

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Sudden infant death with dysgenesis of the testes syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1837371/>

Other Diagnosis and Management Resources

- Clinic for Special Children (Strasburg, Pennsylvania)
<https://clinicforspecialchildren.org/>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Sudden Infant Death Syndrome
<https://medlineplus.gov/suddeninfantdeathsyndrome.html>

Genetic and Rare Diseases Information Center

- Sudden infant death with dysgenesis of the testes syndrome
<https://rarediseases.info.nih.gov/diseases/12382/sudden-infant-death-with-dysgenesis-of-the-testes-syndrome>

Educational Resources

- Disease InfoSearch: Sudden infant death with dysgenesis of the testes syndrome
<http://www.diseaseinfosearch.org/Sudden+infant+death+with+dysgenesis+of+the+testes+syndrome/9364>
- MalaCards: sudden infant death with dysgenesis of the testes syndrome
http://www.malacards.org/card/sudden_infant_death_with_dysgenesis_of_the_testes_syndrome
- Orphanet: Sudden infant death-dysgenesis of the testes syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=168593

Patient Support and Advocacy Resources

- C.J. First Candle
<http://cjfirstcandle.org/>
- Share Pregnancy & Infant Loss Support
<http://nationalshare.org/>
- The Compassionate Friends
<https://www.compassionatefriends.org/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28sudden+infant+death%5BTIAB%5D%29+AND+%28testes%5BTIAB%5D%29+AND+%28dysgenesis%5BTIAB%5D%29%29+OR+%28SIDDT%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- SUDDEN INFANT DEATH WITH DYSGENESIS OF THE TESTES SYNDROME
<http://omim.org/entry/608800>

Sources for This Summary

- Hering R, Frade-Martinez R, Bajanowski T, Poets CF, Tschentscher F, Riess O. Genetic investigation of the TSPYL1 gene in sudden infant death syndrome. *Genet Med*. 2006 Jan;8(1): 55-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16418600>
- Puffenberger EG, Hu-Lince D, Parod JM, Craig DW, Dobrin SE, Conway AR, Donarum EA, Strauss KA, Dunckley T, Cardenas JF, Melmed KR, Wright CA, Liang W, Stafford P, Flynn CR, Morton DH, Stephan DA. Mapping of sudden infant death with dysgenesis of the testes syndrome (SIDDT) by a SNP genome scan and identification of TSPYL loss of function. *Proc Natl Acad Sci U S A*. 2004 Aug 10;101(32):11689-94. Epub 2004 Jul 23.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15273283>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC511011/>
- OMIM: SUDDEN INFANT DEATH WITH DYSGENESIS OF THE TESTES SYNDROME
<http://omim.org/entry/608800>
- Schubert S, Haas C, Bartsch C, Mirshekarnejad M, Kohrs S, Roettinger I, Grosshennig A, Stuhmann M, Scholz C, Schmidtke J. Variants in TSPYL1 are not associated with sudden infant death syndrome in a cohort of deceased infants from Switzerland. *Mol Cell Probes*. 2015 Feb;29(1): 31-4. doi: 10.1016/j.mcp.2014.10.006. Epub 2014 Nov 4.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25449952>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/sudden-infant-death-with-dysgenesis-of-the-testes-syndrome>

Reviewed: December 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services